POLR1C gene

RNA polymerase I subunit C

Normal Function

The *POLR1C* gene provides instructions for making one part (subunit) of two related enzymes called RNA polymerase I and RNA polymerase III. These enzymes are involved in the production (synthesis) of ribonucleic acid (RNA), a chemical cousin of DNA. Both enzymes help synthesize a form of RNA known as ribosomal RNA (rRNA). RNA polymerase III also plays a role in the synthesis of several other forms of RNA, including transfer RNA (tRNA). Ribosomal RNA and transfer RNA assemble protein building blocks (amino acids) into functioning proteins, which is essential for the normal functioning and survival of cells.

Based on its involvement in Treacher Collins syndrome, the *POLR1C* gene appears to play a critical role in the early development of structures that become bones and other tissues of the face.

Health Conditions Related to Genetic Changes

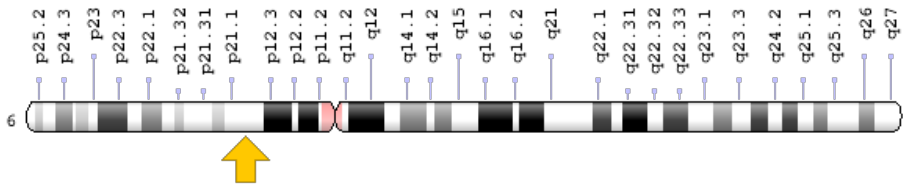
Treacher Collins syndrome

At least six mutations in the *POLR1C* gene have been identified in people with Treacher Collins syndrome, a condition that affects the development of bones and other tissues of the face. These mutations appear to alter the structure and function of the POLR1C protein, which reduces the amount of functional RNA polymerase I and RNA polymerase III in cells. Consequently, less rRNA is produced. Researchers speculate that a shortage of rRNA may trigger the self-destruction (apoptosis) of certain cells involved in the early development of facial bones and tissues. The abnormal cell death could underlie the specific problems with facial development found in Treacher Collins syndrome. However, it is unclear why the effects of a reduction in rRNA are limited to facial development.

Chromosomal Location

Cytogenetic Location: 6p21.1, which is the short (p) arm of chromosome 6 at position 21.1

Molecular Location: base pairs 43,517,024 to 43,562,404 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AC40
- DNA-directed RNA polymerase I subunit C
- DNA-directed RNA polymerases I and III 40 kDa polypeptide
- DNA-directed RNA polymerases I and III subunit RPAC1
- polymerase (RNA) I polypeptide C
- polymerase (RNA) I polypeptide C, 30kDa
- polymerase (RNA) I subunit C
- RNA polymerases I and III subunit AC1
- RPA5
- RPA39
- RPA40
- RPAC1
- RPAC1_HUMAN
- RPC40

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): RNA in Eukaryotic Cells Is Synthesized by Three Types of RNA Polymerase
  https://www.ncbi.nlm.nih.gov/books/NBK22433/#A3981
- Molecular Cell Biology (fourth edition, 2000): The Three Roles of RNA in Protein Synthesis
  https://www.ncbi.nlm.nih.gov/books/NBK21603/
- The Cell: A Molecular Approach (second edition, 2000): Eukaryotic RNA Polymerases
  https://www.ncbi.nlm.nih.gov/books/NBK9935/#A981

GeneReviews

- Treacher Collins Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1532

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28POLR1C%5BTIAB%5D%29+OR+%28%28RPA5%5BTIAB%5D%29+OR+%28TCS3%5BTIAB%5D%29+OR+%28RPA40%5BTIAB%5D%29+OR+%28RPAC1%5BTIAB%5D%29+OR+%28AC40%5BTIAB%5D%29+OR+%28RPC40%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

OMIM

- POLYMERASE I, RNA, SUBUNIT C
  http://omim.org/entry/610060

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=POLR1C%5Bgene%5D
- HGNC Gene Family: RNA polymerase subunits
  http://www.genenames.org/cgi-bin/genefamilies/set/726
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=20194

- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/9533
- UniProt
  http://www.uniprot.org/uniprot/O15160

Sources for This Summary

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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/21261602
- GeneReview: Treacher Collins Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1532
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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9653160
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC20949/

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